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Few People with Cancer Undergo Testing for Inherited Gene Mutations

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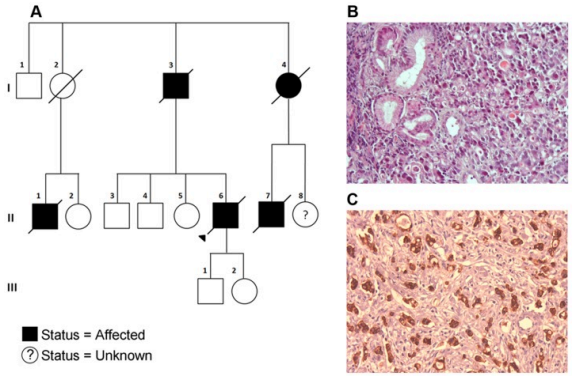
Only a small percentage of people diagnosed with cancer undergo a type of genetic testing, called germline testing, that is highly recommended for those diagnosed with certain cancers. That is the latest finding from an ongoing NCI-funded study.



(A) A family pedigree showing a germline mutation associated with familial gastric cancer. Deceased individuals are marked with a strike-through. (B and C) Pathology images of gastric tumor cells.
Credit: PLoS Genetics. doi: 10.1371/journal.pgen.1004669. (CC BY 4.0)

Tests that look for specific genetic changes in a patient's tumor can help oncologists choose which treatments may be most effective for that individual. Such tests, often called biomarker tests, are widely used in everyday cancer care.

However, guidelines from several medical organizations also recommend that people diagnosed with a host of different cancers also undergo germline testing. These tests use blood samples to look for changes in specific cancer-related genes inherited from a parent.

Like biomarker tests, germline testing can help doctors determine the best treatments for patients, but such testing may also help identify people whose family members should be offered testing for potential cancer-causing gene changes.

Guidelines recommend that germline testing be offered to all people with male breast cancer, ovarian cancer, pancreatic cancer, and metastatic prostate cancer. For other cancers with lower likelihood of harmful inherited mutations, recommendations for germline testing vary.

But new findings from a study that is examining the extent of testing for germline mutations among people diagnosed with cancer in California and Georgia between 2013 and 2019 found that germline testing rates are still low. Among the more than 1.3 million people in the study,

only about 93,000, or 6.8%, underwent germline genetic testing through March 31, 2021, according to findings published July 3 in *JAMA*.

The prevalence of germline testing varied by cancer type, however. Men diagnosed with breast cancer had the highest rate of testing, at 50%. Only about 38% of people with ovarian cancer were tested. Guidelines recommend all people diagnosed with these two cancers receive germline testing. Among women with breast cancer, 26% received testing.

Testing among people with other cancers possibly associated with germline gene mutations was much less common. About 6% of people with endometrial cancer received germline tests, while about 5.6% of people with colorectal and pancreatic cancer received testing. For prostate and lung cancers, the rates of testing were 1.1% and 0.3%.

In an accompanying editorial in *JAMA*, Zsofia Stadler, M.D., and Deborah Schrag, M.D., of Memorial Sloan Kettering Cancer Center, cautioned that "the low rates of cancer genetic testing reported [in the article] raise concern and should stimulate interventions to increase rates of genetic testing with the goal of reducing [the] cancer burden."

Despite improvements, low testing rates still a major concern

To conduct the study, Allison Kurian, M.D., of the Stanford University School of Medicine, and colleagues from several other institutions linked cancer incidence data from the California and Georgia statewide cancer registries—part of NCI's Surveillance, Epidemiology, and End Results (SEER) program—to genetic testing results from the four laboratories that conduct most germline genetic testing in the two states.

These new findings follow up on an earlier study by some of the same researchers who analyzed testing rates in the early 2010s among people with breast and ovarian cancer. That analysis showed low rates of germline testing in people with these two cancers and that, in a small percentage of those who were tested, the results could have warranted changes in their treatment.

As this more recent, superseding analysis found, germline testing for these cancers has increased over time, with testing for people with female breast cancer rising from 24% to 26%. Testing among those with ovarian cancer increased more sharply, with 38.6% being tested by the end of the most recent analysis compared with 30.1% in 2013 and 2014.

Like breast and ovarian cancer, there was also a sharp increase in germline testing among people diagnosed with pancreatic cancer, rising from 1.2% in 2013 to more than 18% in 2019.

Even still, Dr. Kurian and her colleagues cautioned that, although "genetic testing rates increased over time, … even in 2021, [rates] were far lower than 100% for specific cancer types … recommended by practice guidelines."

Disparities in cancer germline testing, and creating new care delivery models

The research team also found substantial disparities in testing rates. Whereas 31% of White people with male or female breast cancer or ovarian cancer underwent germline testing, only 22% to 25% of Asian, Hispanic, and Black people with these cancers had testing.

Drs. Stadler and Schrag pointed out that uncertain germline test results, which are those that identify genetic variants whose effect on cancer risk is not known, "were significantly more common in non-White patients."

That's an important finding, because uncertain results can lead to "suboptimal" patient care and "increased patient anxiety," they wrote, adding that "the combination of low genetic testing and more frequent identification of variants of uncertain clinical significance can perpetuate existing disparities."

Although the study could not account for why germline testing rates remain low, Drs. Stadler and Schrag explained that both health care system related issues, such as limited access to genetic counselors, and patient-level factors, such as uncertainty or mistrust of genetic testing, are likely both key contributors.

"New care delivery models are needed to improve the rates of cancer susceptibility genetic testing," they wrote. The development and testing of such models is a major goal of the Cancer Moonshot℠, they noted.

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